



D2HGDH gene

D-2-hydroxyglutarate dehydrogenase

Normal Function

The *D2HGDH* gene provides instructions for making an enzyme called D-2-hydroxyglutarate dehydrogenase. This enzyme is found in mitochondria, which are the energy-producing centers within cells. Within mitochondria, the enzyme participates in reactions that produce energy for cell activities. Specifically, D-2-hydroxyglutarate dehydrogenase converts a compound called D-2-hydroxyglutarate to another compound called 2-ketoglutarate. A series of additional enzymes further process 2-ketoglutarate to produce energy.

Health Conditions Related to Genetic Changes

2-hydroxyglutaric aciduria

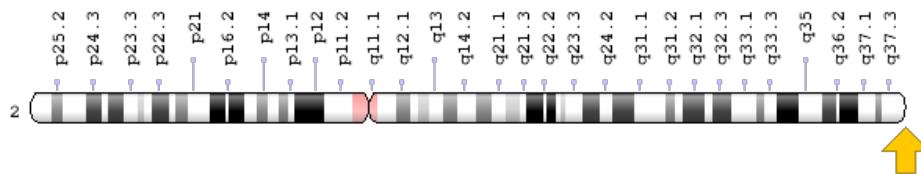
Researchers have identified more than 30 *D2HGDH* gene mutations that cause a type of 2-hydroxyglutaric aciduria known as D-2-hydroxyglutaric aciduria (D-2-HGA) type I. This condition has a variety of signs and symptoms that result primarily from progressive damage to the brain beginning early in life.

Some *D2HGDH* gene mutations change single protein building blocks (amino acids) in the D-2-hydroxyglutarate dehydrogenase enzyme, which likely impairs its function. Other mutations lead to the production of an abnormally short, nonfunctional version of the enzyme. With a shortage of functional enzyme, D-2-hydroxyglutarate is not broken down but instead builds up in cells. At high levels, this compound can damage cells and lead to cell death. Brain cells appear to be the most vulnerable to the toxic effects of this compound, which may explain why the signs and symptoms of D-2-HGA type I primarily involve the brain.

Chromosomal Location

Cytogenetic Location: 2q37.3, which is the long (q) arm of chromosome 2 at position 37.3

Molecular Location: base pairs 241,734,579 to 241,768,816 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 2-hydroxypentanedioic acid
- D2HDH_HUMAN
- D2HGD
- FLJ42195
- MGC25181

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Mitochondrion
<https://www.ncbi.nlm.nih.gov/books/NBK26894/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28D2HGDH%5BTIAB%5D%29+OR+%28D-2-hydroxyglutarate+dehydrogenase%5BTIAB%5D%29+OR+%28D-2-HGDH%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- D-2-HYDROXYGLUTARATE DEHYDROGENASE
<http://omim.org/entry/609186>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
http://atlasgeneticsoncology.org/Genes/GC_D2HGDH.html
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=D2HGDH%5Bgene%5D>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=28358
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/728294>
- **UniProt**
<http://www.uniprot.org/uniprot/Q8N465>

Sources for This Summary

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